
Rocky Hill, CT, 1/21/2021. The Connecticut Department of Public Health's Newborn Screening Program (CT NBS) announced today that screening for Pompe Disease and Mucopolysaccharidosis Type 1 (MPS-I) was implemented on January 1, 2021. Pompe and MPS-I are rare diseases, each caused by a change in a single gene.

As many as four people out of every 100,000 have Pompe Disease. People with Pompe Disease do not have enough of the GAA enzyme that helps the body break down stored sugar. There are two types of Pompe Disease, classical infantile and non-classical late onset. Both can cause muscle problems, heart and lung problems. The classical infantile form begins in early infancy and is the most severe. The non-classical late onset type varies in severity. Problems resulting from the disease can worsen quickly and cause death within the first year. While there is no cure for Pompe, early diagnosis and treatment, including enzyme therapy, can stop symptoms from getting worse. Infants with Pompe disease look healthy when they are born and Newborn Screening is important tool to help to identify newborns with Pompe, so that treatment can start right away.

MPS-I affects one out of every 100,000 people. People with MPS-I do not have enough of the IDUA enzyme that helps the cells break down certain waste products. There are two types of MPS-I, the severe type and the attenuated type. The severe form of MPS-I can cause problems with the heart, airways, eyes and ears, muscles, bones, joints, and brain and can lead to death. Early identification of MPS-I through newborn screening allows for early monitoring and treatment when necessary. Not all children with MPS-I require treatment, but for those who do enzyme replacement therapy and bone marrow transplant can stop problems from getting worse.

The federal Advisory Committee on Heritable Disorders in Newborns and Children recommends screening for Pompe and MPS-I as part of state universal newborn screening (NBS) programs.

The Recommended Uniform Screening Panel (RUSP) is a list of disorders that the Secretary of the Department of Health and Human Services (HHS) recommends for states to screen as part of their state universal newborn screening (NBS) programs. Disorders on the RUSP are chosen based on evidence that supports the potential net benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments. It is recommended that every newborn be screened for all disorders on the RUSP¹.

The CT NBS Program works to ensure that every newborn, who is born or resides in Connecticut, has a valid newborn screening on record and that those infants with abnormal screening results are promptly referred to a specialty treatment center for further evaluation and treatment when needed. These comprehensive efforts help prevent unnecessary disability and premature death. A small amount of blood is taken from the newborn's heel shortly after birth and submitted to the state laboratory to be screened for over 60 conditions.

When a newborn is identified through NBS at being at risk of having Pompe or MPS-I, the CT NBS Program will quickly report the result to the Connecticut Newborn Diagnostic and Treatment Network which works to immediately connect the infant, family and the infant's primary care provider with a specialist with expertise in these disorders to either rule out or confirm the disorder.

Currently, 25 states screen for Pompe and 23 states screen for MPS-I

For more information on Pompe Disease go to <https://www.babysfirsttest.org/newborn-screening/conditions/pompe>

For more information about MPS-I go to <https://www.babysfirsttest.org/newborn-screening/conditions/mucopolysaccharidosis-type-i>

¹ <https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html>

For more information about the CT NBS Program go to <https://portal.ct.gov/newbornscreening>